

# Complex Inheritance and Human Heredity

## section 1 Basic Patterns of Human Inheritance

### ● Before You Read

A family tree shows how people in a family are related. On the lines below, list people who might appear in a family tree. Then read the section to learn how scientists trace inheritance through several generations of a family.

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### MAIN Idea

The inheritance of a trait over several generations is shown in a pedigree.

### What You'll Learn

- how to determine if an inherited trait is dominant or recessive
- examples of dominant and recessive disorders

### ● Read to Learn

#### Recessive Genetic Disorders

During the early 1900s, Gregor Mendel's work on heredity was rediscovered. Archibald Garrod, an English doctor, was studying a disorder that results in black urine and affects bones and joints. Dr. Garrod, with the help of other scientists, discovered that the disorder was a recessive genetic disorder. This finding began the study of human genetics.

Review the table below and recall that a recessive trait is expressed when the person is homozygous recessive for that trait. A person with at least one dominant allele will not express the recessive trait. A person who is heterozygous for a recessive disorder is called a **carrier**.

Term	Description
Homozygous	An organism with two of the same alleles for a particular trait is said to be homozygous for that trait.
Heterozygous	An organism with two different alleles for a particular trait is said to be heterozygous for that trait. When alleles are present in the heterozygous state, the dominant trait will be observed.

### Study Coach

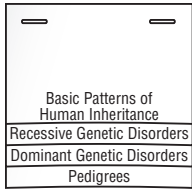
**Create a Quiz** After you read this section, create a quiz based on what you have learned. Then be sure to answer the quiz questions.

### Picture This

1. **Identify** Circle the term that describes the genotype of a person who expresses a recessive trait.



**Take Notes** Use two sheets of paper to make a layered Foldable, as shown below. As you read, record important information you learn about human inheritance.



**Reading Check**

**2. Explain** Why do gangliosides build up in the brain of people with Tay-Sachs disease?

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### What is cystic fibrosis?

Cystic fibrosis is a recessive genetic trait. Chloride ions are not absorbed into cells but are excreted in sweat. Without the chloride ions in cells, water does not diffuse from cells. This causes the secretion of a thick mucus that affects many areas of the body. The mucus interferes with digestion, clogs ducts in the pancreas, and blocks air pathways in the lungs. Patients with cystic fibrosis often get infections because of excess mucus in their lungs.

Treatment includes physical therapy, medicine, special diets, and replacement digestive enzymes. Genetic tests are used to determine if the recessive gene is present.

### What causes albinism?

Albinism is a recessive disorder found in people and animals. In humans, it is caused by the absence of the skin pigment melanin in hair and eyes. People with albinism have white hair, pale skin, and pink eyes. They need to protect their skin from the Sun's ultraviolet rays.

### What is Tay-Sachs disease?

Tay-Sachs (TAY saks) disease is a recessive genetic disorder. Tay-Sachs disease (TSD) is more common among Jews whose ancestors are from eastern Europe.

People with TSD are missing an enzyme needed to break down fatty acids called gangliosides. Normally, gangliosides are made and then destroyed as the brain develops. In people with TSD, gangliosides build up in the brain, causing mental deterioration. Children born with TSD usually die by age five. Currently, there is no cure.

### What causes galactosemia?

Galactosemia (guh lak tuh SEE mee uh) is a recessive genetic disorder. It causes intolerance of the sugar galactose. Milk contains the sugar lactose. During digestion, lactose breaks down into galactose and glucose, the sugar used by the body for energy. People with galactosemia lack the enzyme needed to break down galactose.

### Dominant Genetic Disorders

Not all genetic disorders are recessive. Some are caused by dominant alleles. People who do not have the disorder are always homozygous recessive, meaning they carry two recessive genes for the trait.

## What happens in Huntington's disease?

Huntington's disease is a dominant genetic disorder that affects the nervous system. It is rare. Symptoms occur when the person is between 30 and 50 years old. Symptoms are gradual loss of brain function, uncontrollable movements, and emotional disturbances. Genetic tests can tell people whether they have the gene for Huntington's disease, but there is currently no treatment or cure.

## What is achondroplasia?

Achondroplasia (a kahn droh PLAY zhee uh) is a dominant genetic disorder that is also known as dwarfism. People with this disorder have a small body size and short limbs. They grow to an adult height of about 1.2 m (4 ft).

About 75 percent of people with achondroplasia have parents of average size. Because the gene is dominant, parents who are average size do not have the gene. Therefore, when average-sized parents have a child with achondroplasia, the condition occurs because of a new mutation.

## Pedigrees

Scientists use a diagram called a **pedigree** to trace inheritance of a trait through several generations. A pedigree uses symbols to illustrate inheritance of the trait.

A sample pedigree is shown in the figure below. In the top row, the two symbols connected by a horizontal line are the parents. Their children are listed below them, oldest to youngest from left to right.

Roman numerals are used to represent generations—I for the parents, II for the children, and so on. Arabic numbers are used to represent the individuals within a generation.

### Key to Symbols

○ Normal female

● Female who expresses the trait being studied

◐ Female who is a carrier for the particular trait

| Generation

— Parents

┌┐ Siblings

□ Normal male

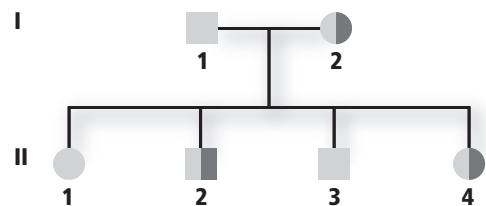
■ Male who expresses the trait being studied

◑ Male who is a carrier for the particular trait

Roman numerals — Generations

Arabic numerals — Individuals in a certain generation

### Example Pedigree



## Think it Over

**3. Explain** How can scientists determine if achondroplasia developed from a new mutation?

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## Picture This

**4. Evaluate** Circle the carriers in the second generation.

## Picture This

**5. Calculate** What percentage of the children in this family inherited Tay-Sachs disease?

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## Picture This

**6. Identify** Do any of the grandchildren in this family have polydactyly?

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## Reading Check

**7. Explain** Why are recessive traits difficult to study?

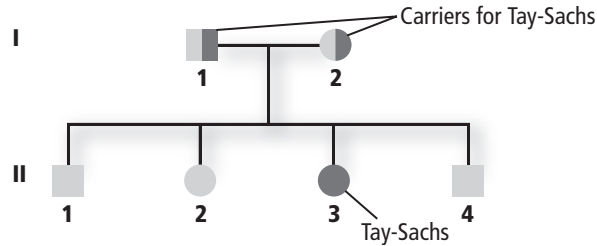
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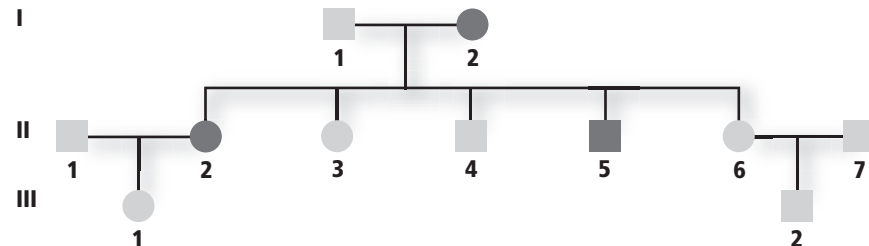
## Analyzing Pedigrees

The figure below is a pedigree showing the inheritance of Tay-Sachs disease, a recessive disorder. The pedigree shows that two parents who do not have Tay-Sachs disease can have a child who has the disorder.



## How is the inheritance of a dominant disorder shown on a pedigree?

The pedigree below shows the inheritance of the dominant disorder, polydactyly (pah lee DAK tuh lee)—extra fingers and toes. A person with dominant disorders could be homozygous or heterozygous for the trait. A person who does not have polydactyly would be homozygous recessive for the trait.



## How are genotypes deduced?

A pedigree can be used to learn the genotype of a person. The genotype is determined by observing the phenotypes, or physical traits, of a person.

Genetic counselors use pedigrees to determine if an inherited trait is dominant or recessive. Dominant traits are easy to recognize. Recessive traits are more difficult because people who carry the allele do not always show the trait.

## Can genetic disorders be predicted?

Scientists can use pedigrees to predict whether a person in a family will get a genetic disorder. Scientists have to follow several people for many generations to accurately study a disorder. Good record keeping within a family can help scientists predict the inheritance of a disorder.